AL-FARABI KAZAKH NATIONAL UNIVERSITY

Faculty of Medicine and Healthcare

Higher School of Medicine

Department of ​Fundamental Medicine

METHODICAL INSTRUCTIONS FOR PRACTICAL CLASSES

Molecular bases of pathology

(medical genetics, general pharmacology)

(10 credits)

Spring semester, 2023-2024 academic year

Medical Genetics

Practical class 1-2

Topic: Introduction to Medical Genetics. Chromosomal disorders.

Content: Classification of Hereditary diseases*.* Chromosomal mutations: characteristics, cause, mechanisms, frequency, phenotypic manifestation, clinical significance. Diagnosis and management of chromosomal disorders. Epidemiology of chromosomal disorders. Down syndrome as an example of autosomal chromosomal diseases (case study): signs and symptoms, causes, mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology

Maximum points – 5

Learning outcomes:

1. draw mind map of hereditary diseases and explain principles of their classification;
2. Remember the structure and function of chromosomes. Compare and contrast their segregation in mitosis and meiosis;
3. Describe the types of structural variation seen in chromosomes (e.g., translocations, inversions, deletions, duplications, copy number variants, etc.). Utilize the presence of a structural variant to explain an individual's risk for a syndrome, reduced fertility, or spontaneous abortion.
4. explain genetic mutations as cause of autosomal chromosomal disorders and summarize their role in clinical variability. Describe how mitotic errors result in mosaicism and explain how it affects the phenotypic expression of genomic disorders;
5. identify clinical manifestation of autosomal chromosomal disorders and explain its clinical variability;
6. compare different diagnostic strategies for diagnosing chromosomal disorders and discuss related ethical and legal issues;
7. compare different management strategies for chromosomal disorders (prevention, treatment) and discuss related ethical and legal issues;
8. summarize epidemiologic data of chromosomal disorders;
9. summarize risk assessment strategy for chromosomal disorders;
10. identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with chromosomal disorders;
11. discuss impact of diagnosis of a genetic condition for the individual and the family;
12. Contrast the uses and limitations of cytogenetic techniques that detect aneuploidy and structural variation including G-banded karyotype, chromosomal microarray analysis (CMA), and fluorescence in situ hybridization (FISH);
13. demonstrate respect of patient’s religious, cultural, social and ethical beliefs and understanding of how that might affect the decisions the patients make;
14. Access reputable resources regarding genetic conditions to inform management and surveillance recommendations. Some resources include: GeneReviews, OMIM, NHGRI Talking Glossary of GeneticTerms, NCBI Genetic Testing Registry, MedlinePlus-Genetics, National Organization for Rare Disorders (NORD), Understanding Rare Chromosome and Gene Disorders (UNIQUE).
15. demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;
16. Describe the basic principles of molecular and cytogenetic nomenclature used to report G-banded karyotype, fluorescence in situ hybridization (FISH), chromosomal microarray analysis (CMA), and sequencing results.

Practical class 3

Topic: Sex Chromosome disorders. Summary of chromosomal diseases.

Content: Classification of Hereditary diseases*.* Chromosomal mutations: characteristics, cause, mechanisms, frequency, phenotypic manifestation, clinical significance. Diagnosis and management of chromosomal disorders. Epidemiology of chromosomal disorders. Turner syndrome as an example of gonosomal chromosomal diseases (case study): signs and symptoms, causes, mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology.

Maximum points – 2.5

Learning outcomes:

1. identify clinical manifestation of gonosomal chromosomal disorders and explain its clinical variability;
2. explain genetic mechanism of gonosomal chromosomal disorders and summarize their role in clinical variability;
3. compare and contrast different diagnostic strategies of gonosomal chromosomal disorders and discuss related ethical and legal issues;
4. compare and contrast different management strategies of gonosomal chromosomal disorders (prevention, treatment) and discuss related ethical and legal issues
5. summarize epidemiologic data of gonosomal chromosomal disorders;
6. summarize risk assessment strategy for gonosomal chromosomal disorders;
7. discus a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with gonosomal chromosomal disorders;
8. Contrast the uses and limitations of molecular techniques that detect nucleotide variants, including Sanger sequencing for single genes, next-generation sequencing for multi-gene, exome, and genome analyses;
9. discuss impact of diagnosis of a genetic condition for the individual and the family;
10. demonstrate respect patient’s religious, cultural, social and ethical beliefs and understanding of how that might affect the decisions the patients make
11. Access reputable resources regarding genetic conditions to inform management and surveillance recommendations. Some resources include: GeneReviews, OMIM, NHGRI Talking Glossary of GeneticTerms, NCBI Genetic Testing Registry, MedlinePlus-Genetics, National Organization for Rare Disorders (NORD), Understanding Rare Chromosome and Gene Disorders (UNIQUE).
12. demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;
13. complete mind map of hereditary diseases and discus principles of their classification;

Practical class 4-5

Topic: Mendelian classic disorders: autosomal inheritance.

Content: Classifications, pathogenetic, pathogenetic mechanisms, epidemiology and management. Classification of classic Mendelian disorders. Gene mutations: characteristics, cause, mechanisms, frequency, phenotypic manifestation, clinical significance. Diagnosis and management of classic Mendelian disorders. Epidemiology of single gene disorders, prognosis. Cystic fibrosis as an example of an autosomal recessive disease (case study): signs and symptoms, causes, mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology

Maximum points – 5

Learning outcomes:

1. draw mind map of single gene diseases (mendelian disorders) and explain principles of their classification;
2. Explain how genetic variation, in coding and non-coding regions of the genome, influences gene expression and can result in disease.
3. Describe the characteristic features of autosomal dominant and autosomal recessive inheritance;
4. identify clinical manifestation of autosomal (dominant and recessive) Mendelian disorders and explain its clinical variability;
5. Describe the ways in which variants occurring within coding regions of the genome affect encoded products (e.g., missense (non-synonymous), nonsense, silent (synonymous), frameshift, and aberrant splicing);
6. Describe the mechanisms by which changes in proteins result in disease (loss-of-function, gain-of-function);
7. Describe the spectrum of genetic liability; from monogenic disorders predominantly caused by single genes with large effect sizes;
8. explain mechanism of autosomal recessive mutations for Cystic fibrosis and summarize their role in clinical variability of it and of autosomal (dominant and recessive) monogenic disorders;
9. compare and contrast different diagnostic strategies of Cystic fibrosis and summarize principles of diagnosis of autosomal (dominant and recessive) monogenic disorders and discuss related ethical and legal issues;
10. compare and contrast different management strategies of Cystic fibrosis and summarize principles of management of autosomal (dominant and recessive) monogenic disorders (prevention, treatment) and discuss related ethical and legal issues;
11. summarize epidemiologic features of autosomal (dominant and recessive) monogenic disorders on the example of Cystic fibrosis;
12. calculate the risk of Cystic fibrosis and summarize risk assessment strategy for autosomal (dominant and recessive) monogenic disorders;
13. identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with autosomal (dominant and recessive) monogenic disorders on the example of Cystic fibrosis;
14. discuss impact of diagnosis of a genetic condition for the individual and the family;
15. demonstrate respect of patient’s religious, cultural, social and ethical beliefs and understanding how that might affect the decisions the patients make;
16. Access reputable resources regarding genetic conditions to inform management and surveillance recommendations. Some resources include: GeneReviews, OMIM, NHGRI Talking Glossary of GeneticTerms, NCBI Genetic Testing Registry, MedlinePlus-Genetics, National Organization for Rare Disorders (NORD), Understanding Rare Chromosome and Gene Disorders (UNIQUE).
17. demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;
18. demonstrate understanding of genetic and medical aspects of chromosomal disorders on colloquium;

Practical class 6

Topic: Mendelian classic disorders: sex-linked inheritance.

Content: Classifications, pathogenetic mechanisms, epidemiology and management. Classification of Mendelian classic disorders. Gene mutations: characteristics, cause, mechanisms, frequency, phenotypic manifestation, clinical significance. Diagnosis and management of Mendelian classic disorders. Epidemiology of single gene disorders, prognosis. Hemophilia as an example of X-linked diseases (case study): signs and symptoms, causes, mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology.

Maximum points – 2.5

Learning outcomes:

1. complete mind map of single gene diseases (Mendelian disorders) and explain principles of their classification;
2. identify clinical features of Hemophilia and explain its clinical variability and summarize clinical manifestation of sex-linked Mendelian disorders;
3. explain mechanism of genetic mutations in case of Hemophilia and summarize their role in clinical variability of sex-linked Mendelian disorders;
4. Describe the characteristic features of X-linked (including manifesting carrier/skewed X-inactivation patterns) and Y-linked inheritance;
5. compare and contrast different diagnostic strategies of Hemophilia and summarize principles of diagnosis of sex-linked Mendelian disorder and discuss related ethical and legal issues;
6. compare and contrast different management strategies of Hemophilia and summarize principles of management of sex-linked Mendelian disorder (prevention, treatment) and discuss related ethical and legal issues;
7. summarize epidemiologic data of sex-linked Mendelian disorders on the example of Hemophilia
8. calculate the risk of Hemophilia and summarize risk assessment strategy for sex-linked Mendelian disorders;
9. identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with sex-linked Mendelian disorders on the example of Hemophilia;
10. discuss impact of diagnosis of a genetic condition on the individual and the family;
11. demonstrate respect of patient’s religious, cultural, social and ethical beliefs and understand how that might affect the decisions the patients make
12. Access reputable resources regarding genetic conditions to inform management and surveillance recommendations. Some resources include: GeneReviews, OMIM, NHGRI Talking Glossary of GeneticTerms, NCBI Genetic Testing Registry, MedlinePlus-Genetics, National Organization for Rare Disorders (NORD), Understanding Rare Chromosome and Gene Disorders (UNIQUE).
13. demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;

Practical class 7

Topic: Biochemical bases of hereditary metabolic disorders

Content: Introduction. Enzymopathies. Classification of enzymopathies: hereditary and acquired. Consequences of enzymatic defects. Enzymopathies in carbohydrate metabolism. Biochemical disorders in enzymopathy in carbohydrate metabolism. Hereditary lactase deficiency. Glycogenoses. Galactosemia. Fructosemia. Mucopolysaccharidoses. Genetics of diabetes mellitus. Diagnostics. Prenatal diagnosis. Screening in newborns. Treatment.

Maximum points – 2.5

Learning outcomes:

1. Describe the molecular basis of metabolic pathways and its significance.
2. Know the basic molecular biochemical mechanisms of the occurrence and development of enzymopathy in carbohydrate metabolism and possible ways of their prevention and diagnosis.
3. Understand the molecular genetic features of the development of diabetes mellitus.
4. Interpret the results of laboratory data - biochemical indicators indicating hereditary carbohydrate metabolism disorders

Practical class 8

Topic: Biochemical disorders in lipid metabolism enzymopathy

Content: Genetic defects in medium chain fatty acid acyl-CoA dehydrogenase. Hereditary hypercholesterolemia. Obesity gene – obese gene. Lysosomal diseases. Hereditary disorders of amino acid metabolism: phenylketonuria; alkaptonuria; alkaptonuria, homocysteinuria, etc. Enzymopathies leading to disruption of urea synthesis. Diagnostics. Treatment.

Maximum points – 2.5

Learning outcomes:

1. Know the basic molecular biochemical mechanisms of the occurrence and development of enzymopathy in lipid and protein metabolism and possible ways of their prevention and diagnosis.
2. Interpret the results of laboratory data - biochemical indicators indicating hereditary disorders of lipid and protein metabolism.

Practical class 9

Topic: Non-mendelian genetic disorders.

Content: Causes, classifications, pathogenetic mechanisms, epidemiology, diagnosis and management. Mitochondrial diseases. Genomic imprinting. Epigenetic of depression. Trinucleotide Repeat disorders. Huntington's Disease as an example of non-mendelian genetic disorders (case study): signs and symptoms, causes, mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology. Summary of Monogenic diseases

Maximum points – 2.5

Learning outcomes:

1. draw mind map of non-mendelian genetic disorders and explain principles of their classification
2. identify clinical features of Huntington’s Disease and explain its clinical variability and summarize clinical manifestation of this disorders;
3. explain mechanism of genetic mutations Huntington's Disease and summarize their role in clinical variability of non-mendelian genetic disorders;
4. Describe the underlying genetic mechanisms of non-Mendelian inheritance patterns: mitochondrial, somatic and germline mosaicism, uniparental disomy, parent of origin (epigenetic and genomic imprinting) and repeat expansion disorders. Explain how these phenomena affect phenotype and recurrence risk;
5. Describe the concept of epigenetics and explain the role of epigenetic mechanisms in the regulation of gene expression and how it can influence disease. Recognize that some epigenetic modifications can change over time.
6. compare and contrast different diagnostic strategies of Huntington's Disease and summarize principles of diagnosis of non-mendelian genetic disorders and discuss related ethical and legal issues;
7. compare and contrast different management strategies of Huntington's Disease and summarize principles of management of non-mendelian genetic disorders (prevention, treatment) and discuss related ethical and legal issues;
8. summarize epidemiologic data of non-mendelian genetic disorders on the example of Huntington's Disease;
9. calculate the risk of Huntington’s Disease and summarize risk assessment strategy for non-mendelian genetic disorders;
10. identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with non-mendelian genetic disorders on the example of Huntington's Disease;
11. discuss impact of diagnosis of a genetic condition on the individual and the family;
12. demonstrate respect of patient’s religious, cultural, social and ethical beliefs and understand how that might affect the decisions the patients make
13. Access reputable resources regarding genetic conditions to inform management and surveillance recommendations. Some resources include: GeneReviews, OMIM, NHGRI Talking Glossary of GeneticTerms, NCBI Genetic Testing Registry, MedlinePlus-Genetics, National Organization for Rare Disorders (NORD), Understanding Rare Chromosome and Gene Disorders (UNIQUE).
14. demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;
15. Explain the concept of genotype-phenotype correlation and how factors such as reduced penetrance (including age-dependent penetrance), variable expressivity, genetic heterogeneity (locus and allelic), pleiotropy, modifier genes, de novo pathogenic variants, and environmental factors affect the phenotypic expression of a disease and the observed pattern of inheritance.

Practical class 10

Topic: Fundamentals of the Population Genetics.

Content: Demographic characteristics, types of populations, marital structure of populations, genetic characteristics of the population. Hardy-Weinberg’s law. Genetic burden of populations: concept and medical significance.

Maximum points – 2.5

Learning outcomes:

1. Compare and contrast genetic and geographic ancestry with the social constructs of race and ethnicity. Give examples of situations in which race, ethnicity, and ancestry correlate with different genetic and environmental risk factors for disease. Explain how these situations may contribute to incorrect inferences about health in individuals and groups;
2. Explain how concepts of population genetics (including population bottlenecks, founder effects, and natural selection) contribute to allele frequency differences across populations. Discuss how these concepts can alter allele frequencies in the population, leading to increased risk in specific populations;
3. Calculate the estimated genotype frequencies within a population using the Hardy-Weinberg equation and describe how they can be used to predict genetic disease risk and carrier status;
4. discuss the importance of determining the burden of hereditary diseases in human populations, the study of the magnitude and structure of the burden of hereditary diseases to determine the amount of medical, social and rehabilitation assistance to the population.

Practical class 11

Topic: Pharmacogenetics.

Content:

Maximum points – 2.5

Learning outcomes:

1. Define pharmacogenetics/genomics and explain how the variants in genes involved in drug transport and metabolism contribute to variability in drug response;

Practical class 12-13

Topic: Polygenic multifactorial disorders.

Content: characteristics, cause, mechanisms, frequency, phenotypic manifestation, clinical significance. Diagnosis and management of polygenic diseases. Epidemiology of polygenic diseases. Diabetes mellitus as an example of polygenic multifactorial disorders (case study): signs and symptoms, classification, causes, mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology. How Insulin works. The role of Glucose. Prediabetes: causes and risk groups. Differences Diabetes 1 type and Diabetes 2 type. Complications.

Maximum points – 5

1. continue work on mind map of hereditary diseases and explain principles of classification of polygenic disorders;
2. identify clinical features of Diabetes mellitus and explain its clinical variability and summarize clinical manifestation of polygenic disorders;
3. Explain the principles of multifactorial inheritance;
4. compare and contrast different strategies in genetic profiling of Diabetes mellitus and summarize principles of diagnosis and genetic screening of polygenic disorders and discuss related ethical and legal issues;
5. compare and contrast different management strategies of Diabetes mellitus and summarize principles of management of polygenic disorders (prevention, treatment) and discuss related ethical and legal issues;
6. summarize epidemiologic data of polygenic disorders on the example of Diabetes mellitus;
7. calculate the risk of Diabetes mellitus and summarize risk assessment strategies for polygenic disorders;
8. identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with polygenic disorders on the example of Diabetes mellitus;
9. discuss impact of diagnosis of a genetic condition on the individual and the family;
10. demonstrate respect of patient’s religious, cultural, social and ethical beliefs and understand how that might affect the decisions the patients make
11. Access reputable resources regarding genetic conditions to inform management and surveillance recommendations. Some resources include: GeneReviews, OMIM, NHGRI Talking Glossary of GeneticTerms, NCBI Genetic Testing Registry, MedlinePlus-Genetics, National Organization for Rare Disorders (NORD), Understanding Rare Chromosome and Gene Disorders (UNIQUE).
12. demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;
13. Describe the principles of genome-wide association studies and how they are used to identify correlations between genomic regions and disease susceptibility. Explain the strengths and limitations of this approach;

Practical class 14

Topic: Cancer Genetics and Genomics.

Content: Cancer genes. Hereditary cancer syndromes. Familial occurrence of cancer. Sporadic cancer and genetic bases. Genetic technology in cancer prevention, diagnosis and therapy. [Breast cancer](https://en.wikipedia.org/wiki/Breast_cancer) (case study): causes and genetic mechanism (clinical and genetic variants), diagnosis, management (prevention and treatment), prognosis, epidemiology.

Maximum points – 2.5

Learning outcomes:

1. continue work on mind map of hereditary diseases and explain principles of their classification
2. identify clinical features of [breast cancer](https://en.wikipedia.org/wiki/Breast_cancer) and explain its lifetime prevalence, genetic variability and summarize data on hereditary cancer syndromes and syndromes with familial cancer;
3. explain genetic mechanisms of oncogenesis on [breast cancer](https://en.wikipedia.org/wiki/Breast_cancer) example and summarize their role in clinical variability of hereditary cancer syndromes and syndromes with familial cancer;
4. compare and contrast different diagnostic strategies of [breast cancer](https://en.wikipedia.org/wiki/Breast_cancer) and summarize principles of diagnosis of hereditary cancer syndromes and syndromes with familial cancer and discuss related ethical and legal issues;
5. compare and contrast different management strategies of [breast cancer](https://en.wikipedia.org/wiki/Breast_cancer) and summarize principles of management of hereditary cancer syndromes and syndromes with familial cancer (prevention, treatment) and discuss related ethical and legal issues;
6. summarize epidemiologic data of hereditary cancer syndromes and syndromes with familial cancer on the example of [breast cancer](https://en.wikipedia.org/wiki/Breast_cancer);
7. calculate the risk of [breast cancer](https://en.wikipedia.org/wiki/Breast_cancer) and summarize risk assessment strategies for hereditary cancer syndromes and syndromes with familial cancer;
8. identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with hereditary cancer syndromes and syndromes with familial cancer on the example of [breast cancer](https://en.wikipedia.org/wiki/Breast_cancer)
9. discuss impact of diagnosis of hereditary cancer syndromes and syndromes with familial cancer on the individual and the family;
10. demonstrate respect of patient’s religious, cultural, social and ethical beliefs and understand how that might affect the decisions the patients make
11. Access reputable resources regarding genetic conditions to inform management and surveillance recommendations. Some resources include: GeneReviews, OMIM, NHGRI Talking Glossary of GeneticTerms, NCBI Genetic Testing Registry, MedlinePlus-Genetics, National Organization for Rare Disorders (NORD), Understanding Rare Chromosome and Gene Disorders (UNIQUE).
12. demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;
13. Describe the multistep model of cancer pathogenesis and the role that DNA repair, proto-oncogenic, and tumor suppressor genes play in this model.
14. Recognize the types of genetic and epigenetic changes that can result in gain-of-function of protooncogenes or loss-of-function of tumor suppressor genes (e.g., Knudson two-hit hypothesis).
15. Explain how cancer is multifactorial in nature and describe the role of different risk factors in cancer development including germline (high, moderate, and low penetrance) pathogenic variants, familial predisposition, environmental factors (smoking, alcohol, diet, estrogen exposure, radiation/UV light), and chance (i.e., sporadic).
16. Explain why germline pathogenic variants associated with hereditary cancer predisposition syndromes are often characterized by earlier onset of cancer, increased risk of multiple cancers in a single individual and patterns of specific cancers in a family.
17. Describe the application of current somatic/tumor and germline testing (cytogenetic, molecular, and epigenetic technologies) to clarify the mechanism of tumorigenesis, evolution, diagnosis, and prognosis of cancer.
18. Describe how genetic testing of the tumor and/or germline can lead to individualized and targeted cancer therapies (precision medicine), and/or long term follow up. Explain the mechanisms of action of these therapies (e.g., Inhibitors of: PARP, PD-1/PD-L1, IDH1/2, tyrosine kinases).
19. Assess the likelihood of a hereditary cancer predisposition by evaluating an individual’s personal and family history of cancer, including early age of onset of cancer, affected family members, multiple primary cancers, and type of cancer (e.g., BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer, Lynch syndrome).
20. Recognize phenotypic characteristics of syndromic conditions with increased cancer risk that may benefit from a genetic evaluation (e.g., Peutz-Jeghers Syndrome, PTEN-Hamartoma Syndrome, Neurofibromatosis Type 1, Tuberous Sclerosis Complex).
21. Recognize the benefits of germline testing in families with hereditary cancer predisposition syndromes including opportunity to improve disease surveillance in genotype positive but asymptomatic family members, provision of surgical and/or other treatment options based on the known patterns and penetrance (high/moderate/low risk) of the cancer type.

Practical class 15

Topic: Metabolic aspects of cancer

Content: Molecular biochemical mechanisms of tumor growth. Mechanisms of neoplastic transformation. Features of tumor cell metabolism. Biochemical markers of tumor cells. Biochemical disturbances in the body that accompany tumor growth. Metabolic immunosuppression. Paraneoplastic endocrine syndromes. Biochemical and molecular biological basis for early diagnosis and chemotherapy of malignant neoplasms. Proteomics and metabolomics. OMICS biomarkers and early diagnosis.

Maximum points – 2.5

Learning outcomes:

1. Describe the features of tumor cell metabolism.
2. Understand the medical aspects of proteomics and modern molecular biology techniques.
3. Interpret the results of laboratory data - biochemical indicators indicating tumor cells

Practical class 16

Topic: Polygenic disorders: developmental malformation.

Content: Classification, cause, mechanisms, prevalence rates, phenotypic manifestation, clinical significance. Neural tube defects (NTDs) as example (case study): definition, types, causes, mechanisms, diagnosis, prevention, epidemiology and management.

Maximum points – 2.5

Learning outcomes:

1. continue work on mind map of hereditary diseases and explain principles of their classification
2. identify clinical features of NTDs and explain its clinical variability and summarize clinical manifestation of developmental malformation in general;
3. explain mechanism of genetic causes of NTDs and summarize their role in clinical variability of developmental malformation in general;
4. compare and contrast different diagnostic strategies of NTDs and summarize principles of diagnosis of developmental malformation in general and discuss related ethical and legal issues;
5. compare and contrast different management strategies of NTDs and summarize principles of management of developmental malformation in general (prevention, treatment) and discuss related ethical and legal issues;
6. summarize epidemiologic data of developmental malformation in general on the example of NTDs;
7. calculate the risk of NTDs and summarize risk assessment strategy for developmental malformation;
8. identify members of a multidisciplinary team to deliver genetic diagnosis and counseling and appropriate genetic support groups for the patients and their family with developmental malformation on the example of NTDs;
9. discuss impact of diagnosis of developmental malformation on the individual and the family;
10. demonstrate respect of patient’s religious, cultural, social and ethical beliefs and understand how that might affect the decisions the patients make;
11. Access reputable resources regarding genetic conditions to inform management and surveillance recommendations. Some resources include: GeneReviews, OMIM, NHGRI Talking Glossary of GeneticTerms, NCBI Genetic Testing Registry, MedlinePlus-Genetics, National Organization for Rare Disorders (NORD), Understanding Rare Chromosome and Gene Disorders (UNIQUE).
12. demonstrate willingness and desire to learning with readiness to listen and learn from peers and patients;

Practical class 17-18

Topic: Genetic counseling. Genetic testing, prevention and treatment.

Content:

Maximum points – 5

Learning outcomes:

1. Explain the rationale for referring a patient for a clinical genetic evaluation based upon medical history and physical exam findings that may include congenital anomalies, neurodevelopmental phenotype, unusual physical features or stature, multisystemic disease, early onset, bilateral or atypical disease, and multiple miscarriages or reproductive failure.
2. Use the family history to evaluate indicators for clinical genetics referral including multiple affected family members with the same or significantly overlapping clinical presentation for example: physical findings, developmental delay/intellectual disabilities, clustering of cancers, multiple miscarriages, fetal or early childhood deaths, or sudden cardiac death.
3. Use a family history to construct a three-generation pedigree and interpret for possible mode(s) of inheritance (Mendelian, multifactorial, and mitochondrial) and assessment of associated recurrence risks. Apply the most up-to-date guidelines to document family history using inclusive practices to indicate gender identity, family structure, use of assisted-reproduction technologies, and/or adoption.
4. Recognize that congenital anomalies may have intrinsic or extrinsic causes, may occur in isolation or part of a pattern, and that these anomalies (malformation, deformation, and disruption) may be part of a syndrome, sequence, and association
5. Explain the difference between screening (i.e., noninvasive prenatal screening (cell-free DNA), newborn screening, carrier screening), and diagnostic or predictive genetic and genomic testing strategies as components in the evaluation of a patient.
6. Discriminate between testing strategies with respect to the type of tissues evaluated (somatic vs. constitutional /germline).
7. Recognize that the optimal approach is to test an affected family member first (informative vs. uninformative testing result) and that predictive genetic testing of unaffected and/or asymptomatic family members has ethical considerations.
8. Select genetic tests most appropriate for a patient's suspected diagnosis. Recognize that testing can result in differences in management (including preventive screening, changes to medication/dosing, surgery).
9. Interpret the results of a cytogenetic (G-banded karyotype, FISH or microarray) report with respect to common numerical and structural chromosome abnormalities, and recognize their clinical features, etiologies and prognoses (e.g. trisomy 13, 18, 21; 47,XXY (Klinefelter syndrome); 45,X (Turner syndrome); 22q11.2 deletion syndrome (DiGeorge syndrome); 7q11.23 deletion (Williams syndrome), deletion 15q11.2 ( Prader-Willi/Angelman syndrome).
10. Recognize the role of biochemical screening studies (e.g., plasma ammonia, plasma acylcarnitine profile, plasma amino acids, and urine organic acids) in the diagnosis of an inborn error of metabolism. Interpret the results in the context of the patient’s clinical presentation.
11. Compare and contrast the value, sensitivity, and specificity of molecular and biochemical testing strategies in the diagnosis and management of metabolic disorders.
12. Recognize the benefits and limitations of direct-to-consumer and consumer-initiated testing and how they may impact the patient experience and their care.
13. Recognize the indications and limitations for carrier screening including test characteristics, carrier frequency, and personal/family history.
14. Discuss benefits, and limitations of currently used prenatal screening approaches, including: first and second trimester serum screening, noninvasive prenatal screening, and ultrasound evaluation.
15. Discuss risks, benefits, and limitations of invasive prenatal diagnostic techniques including chorionic villi sampling, amniocentesis, and cordocentesis.
16. Discuss indications for preimplantation genetic testing.
17. Recognize the impact of teratogens (e.g. alcohol, drugs, infectious agents, hyperglycemia secondary to maternal diabetes) on embryonic development, including the effect of dosage, timing and duration of exposure.
18. Outline the principles for management of genetic diseases and describe their appropriate application based on disease pathogenesis. Examples include correction, enhancement or replacement of a defective structural protein or enzyme, dietary treatment, modulation of RNA expression or function, alteration of DNA sequence to modulate gene expression using techniques such as genome editing for somatic and germline therapies, organ transplantation, and stem cell therapy.
19. Define gene therapy and explain the current techniques. Outline scientific, ethical, and clinical obstacles to widespread implementation of gene therapy.
20. Recognize how identification of a specific pathogenic variant in an individual could lead to targeted treatment and/or management of disease. Some examples include: BCR-ABL1 fusion (tyrosine kinase inhibitors such as imantinib), PML-RARA fusion (all-trans retinoic acid (ATRA)), exon amenable skipping for Duchenne muscular dystrophy (DMD, exon 51- eteplirsen), cystic fibrosis (CFTR, G551D- ivacaftor).
21. Explain how the presence of specific genetic variants that affect drug transport or metabolism in an individual patient (pharmacogenetics) may predict physiological response or adverse reactions to medications and influence medical management. Some examples include: CYP2C19 (clopidogrel), CYP2C9 (warfarin), HLAB\*1502 (carbamazepine). Refer to PharmGKB.org as a good resource for drug specific information.
22. Recognize the emerging role of polygenic risk scores in complex conditions and describe how modifications of lifestyle and environment can prevent or mitigate disease in genetically predisposed individuals. Recognize that there may be limited portability of polygenic risk scores developed using data from individuals of one ancestry to individuals of other ancestries.
23. Address fears and concerns of patients and family members regarding genetic testing in order to enable informed decision-making in a respectful, sensitive, and non-directive manner.
24. Describe the role of clinical genetics professionals (e.g., medical geneticists, genetic counselors, clinical laboratory geneticists) in patient care and the process for making appropriate referrals for genetic evaluations.
25. Demonstrate the ability to explain to patients and families the rationale for a genetic evaluation and basic concepts of inheritance.
26. Explain to the patient/family the rationale behind genetic evaluation and the components of genetic testing as they relate to informed consent. These components include potential risks, benefits, and limitations of different types of results (i.e., diagnostic, non-diagnostic, variants of uncertain clinical significance, and secondary findings).
27. Recognize the relevance of patient autonomy in the informed consent/assent for genetic testing of minors (i.e., the unique implication of testing the minor for an adult-onset disorder).
28. Communicate family history and medical history pertinent to genetics with an interdisciplinary team of healthcare professionals.
29. Evaluate personal knowledge and expertise about genetic counseling and testing to decide if and when to initiate pre-test counseling and genetic testing or to refer for specialty services.
30. Explain how a patient’s autonomy (including their own privacy and confidentiality) can conflict with the healthcare provider’s duty to warn potentially at-risk family members about genetic health risk.
31. Demonstrate compassion and respect regarding the potential socio-emotional impact of disclosing predictive genetic testing results to patients and families.
32. Validate the individual identity of patients by recognizing how their unique characteristics may influence their decisions regarding genetic testing and management.
33. Explain the rationale for inclusion of a disease in a newborn screening program and the criteria that are important for a successful genetic screening program.
34. Describe ethical challenges related to genetic information, including ensuring privacy and potential discrimination, and the ways in which the Genetic Information Non-Discrimination Act (GINA) aims to address some of these issues.
35. Identify historical examples of how incorrect assumptions about correlations between the social construct of race and genetic ancestry have been used to justify inequitable and unjust policies and practices in health care and society more widely. Propose ways to minimize these harms in the present and future.

METHODICAL INSTRUCTIONS FOR PRACTICAL CLASSES

Aim: to enforce understanding of pathogenesis, methods of diagnosis and management of genetically determined and hereditary diseases, develop problem solving, team-working and self-learning skills.

Learning outcomes:

1. *apply knowledge about molecular and genetic aspects of genetically determined diseases (chromosomal, monogenic, polygenic); understand the principles of genetic diagnostics and medical genetic counseling.*
2. *understand the biochemical processes in the main pathological conditions and genetically determined diseases.*
3. *interpret the results of specific molecular genetic diagnostic methods*
4. *understand the role of relevant risk factors of diseases for decision-making with a view to their prevention.*
5. *integrate knowledge on human genetics for the purposes of diagnosis and personalized treatment of human pathology*
6. *demonstrate the ability to identify learning gaps and create strategies to enhance one’s own knowledge and skills.*
7. *effectively communicate with other students and teachers regarding medical and scientific information, articulate their opinions clearly when discussing and work effectively as a member of the team.*

Work schedule

1. Familiarize yourself with the basic and additional literature, use textbooks, the syllabus and present directions, Internet resources to prepare for seminars.

2. Be prepared for class and participate actively on case-discussion and problem-solving group activities.

3. Use the examples (in this number cases and your own experience studied before) for illustration of theoretic material.

4. Use different tools for studying, discussion and visualization of thoughts - drawing, mind maps, 3d-modelling.

5. Use the group work with cases for the development of teamwork skills, communication, problem solving and self-studying.

General Pharmacology

Practical class 1

Topic: Introduction to Pharmacology.The value of the subject. Dosage Forms. INN, trade names. Medicinal dosage forms. Drug prescription.

Maximum points – 2.5

Learning outcomes:

1. *Explain the purpose of the science of pharmacology and its basic terms.*
2. *List the basic dosage forms.*
3. *Explain the principles of naming drugs (Chemical names, international nonproprietary name, trade names, original and generic)*
4. *Write a prescription for the drug.*

Practical class 2

Topic: Pharmacokinetics. Principles of interaction of human bodies with the drugs. Absorption, distribution, biotransformation and excretion of chemicals. Effects of impaired organ functions on pharmacokinetics.

Maximum points – 2.5

Learning outcomes:

1. *Explain terms: “pharmacokinetics, absorption, distribution, biotransformation, excretion”*
2. *Recognize the routes of drugs inside human bodies*
3. *Apply this knowledge when describing a drug.*

Practical class 3

Topic: Pharmacodynamics.Principles of interaction of drugs with human bodies. Different mechanisms of action – agonism and antagonism to different types and subtypes of receptors, inhibition of enzymes, blocking or opening of channels.

Maximum points – 2.5

Learning Outcomes:

1. *Explain terms: “pharmacodynamics, receptor, channel, enzyme, agonist, antagonist, partial agonist, inhibitor, channel blocker, channel transporter”*
2. *Understand the mechanism of drug action on chemical and anatomical basis.*
3. *Apply this knowledge when describing a drug*

Practical class 4

Topic: PNS. Cholinergic drugs.Acetylcholine, it’s function in healthy human body. M and N cholinoreceptors, different subtypes. Cholinomimetics. Cholinesterase inhibitors.

Maximum points – 2.5

Learning Outcomes:

1. *Explain the functions and location of M1, M2, M3, NN, NM receptors throughout the human body.*
2. *Describe action of cholinesterase inhibitors.*
3. *Demonstrate, how selectivity to different types of receptors linked to drug action.*
4. *Characterize (indications, contraindications, side effects) of this drugs: Pilocarpine, Physostigmine, Galantamine, Neostigmine, Nicotine, Cytisine.*

Practical class 5

Topic: PNS. Cholinergic drugs. Cholinoblockers. Cholinesterase reactivators.

Maximum points – 2.5

Learning Outcomes:

1. *Explain the main effects of cholinoblockers.*
2. *Describe action of cholinoblockers and cholinesterase reactivators.*
3. *Demonstrate, how selectivity to different types of receptors linked to drug action.*
4. *Characterize (indications, contraindications, side effects) of this drugs: Pipekuronium, Succinylcholine, Atropine, Solifenacin. Hyoscine, Piridoxim*

Practical class 6

Topic: PNS. Adrenergic drugs. Noradrenaline and adrenaline (Norepinephrine and epinephrine), their functions in healthy human body. Alfa and beta adrenoreceptors, different subtypes. Adrenomimetics.

Maximum points – 2.5

Learning Outcomes:

* + - 1. *Describe the functions and location of α1, α2, β1, β2, β3 receptor subtypes throughout the human body.*
      2. *Describe action of adrenomimetics.*
      3. *Demonstrate, how selectivity to different types of receptors linked to drug action.*
      4. *Characterize (indications, contraindications, side effects) this drugs: Adrenaline (epinephrine), Phenylephrine, Naphazoline, Ephedrine, Clonidine, Dobutamine, Salbutamol, Salmeterol, Isoprenaline (historical)*

Practical class 7

Topic: PNS. Adrenergic drugs. Adrenoblockers, their functions in healthy human body. Alfa and beta adrenoreceptors blockers, different subtypes. Sympatholytics.

Maximum points – 2.5

Learning Outcomes:

*Describe action of adrenoblockers.*

*Demonstrate, how selectivity to different types of receptors linked to drug action.*

*Characterize (indications, contraindications, side effects) this drugs:Phentolamine (historical), Yohimbine, Prazosin / Doxazosin, Propranolol, Metoprolol, Labetalol, Carvedilol, Atenolol.*

Practical class 8

Topic: Hypnotics

Maximum points – 2.5

Learning Outcomes:

1. *Explain mechanisms of Hypnotics.*
2. *Explain function of Hypnotics.*
3. *Explain mechanisms of regulation of GABA.*
4. *Compare different Hypnotics.*

Practical class 9

Topic: *Local Anesthetics*

Maximum points – 2.5

Learning Outcomes:

1. *Explain mechanisms of Local Anesthetics.*
2. *Explain function of Nociceptors.*
3. *Explain mechanisms of regulation of pain.*
4. *Compare different Local Anesthetics*

Practical class 10

Topic: *Anti allergics SAIDS*

Maximum points – 2.5

Learning Outcomes:

1. *Explain mechanisms of allergy.*
2. *Explain function of Hystamin Bradikinin.*
3. *Explain mechanisms of inflamation.*
4. *Compare different Local Anesthetics.*

Practical class 11

Topic: Antianginal drugs. Antihypertensive drugs. Diuretics, Ca channel blockers, Nitrates, ACEI

Maximum points – 2.5

Learning Outcomes:

1. *Explain mechanisms of cardiac ischemia.*
2. *Explain function of RAAS (renin-angiotensin-aldosterone system).*
3. *Explain mechanisms of regulation of water-salt balance.*
4. *Compare different antianginal drugs.*
5. *Characterize (indications, contraindications, side effects) these drugs: Nitroglycerin, Isosorbite dinitrate, verapamil, alpha-blockers (repeat), Beta-blockers (repeat), captopril, enalapril, losartan, nifedipine, amlodipine, clonidine, moxonidine, furosemide, hydrochlorothiazide, indapamide, spironolactone.*

Practical class 12

Topic: Pharmacology of the hematopoietic system and hemostasis.Preparations for the treatment of anemia.Coagulation disorders. Drugs, enhancing drugs and reducing coagulation.Drugs, increasing and reducing platelet aggregation.

Maximum points – 2.5

Learning Outcomes:

1. *Compare and define different causes of anemia*

Practical class 13

Topic: Pharmacology of ES. Diabetes melitus.

Maximum points – 2.5

Learning Outcomes:

*1.* *Compare mechanisms of development of type I and type II diabetes melitus.*

*2. Explain principal insulin replacement therapy, its principles.*

*3. Describe drugs, used in in treatment of type II diabetes: Insulin secretagogues. Insulin sensitizers. Agents acting on the absorption and excretion of glucose.*

*4. Tell the function of glucagon and amylin*

*5. Characterize (indications, contraindications, side effects) these drugs: insulins, metformin, glibenclamide, repaglinide, pioglitazone, canagliflozin, liraglutide, sitagliptin*

Practical class 14

Topic: Anti-inflammatory drugs. Nonsteroidal anti-inflammatory drugs. Steroidal anti-inflammatory drugs.

Maximum points – 2.5

Learning Outcomes:

1. *Compare the functions and location of COX-1 and COX-2 enzyme subtypes throughout the human body.*
2. *List hormones f adrenal cortex*
3. *Characterize (indications, contraindications, side effects) these drugs: aspirin, diclofenac, ibuprofen, celecoxib, meloxicam* *prednisolone, dexamethasone, fludrocortisone.*

Practical class 15

Topic: Opioid system. Opioid agonists and antagonists. Drug addiction

Maximum points – 2.5

Learning Outcomes:

1. *Explain functions of nociceptive and antinociceptive system.*
2. *Tell about opioid receptors, their agonists and antagonists.*
3. *Give definitions to “abuse”, “addiction”, “tolerance”, “dependence”, withdrawal”*
4. *Compare physical and psychological dependence*
5. *Characterize (indications, contraindications, side effects) these drugs: morphine, fentanyl, tramadol, buprenorphine, naloxone.*

Practical class 16

Topic: Antibiotics. Principles of antimicrobial therapy. Mechanisms of formation, prevention and overcoming of resistance. Beta-lactams, Macrolides, Tetracyclines, Peptide antibiotics. Aminoglycosides.

Maximum points – 2.5

Learning Outcomes:

1. *Explain the difference between gram-positive and gram-negative bacteria*
2. *Define fungi, chlamydia, mycoplasma, viruses.*
3. *Explain mechanisms of development of resistance*
4. *List methods of overcoming resistance*
5. *Characterize (indications, contraindications, side effects) this drugs: Penicillin, amoxicillin, oxacillin, cefazolin, cefuroxime, ceftriaxone cefepime, Ceftaroline, imipenem, aztreonam, clindamycin, erythromycin, azithromycin, clarithromycin, Streptomycin, gentamicin, doxycycline, Tigecycline, chloramphenicol,**vancomycin,*

Practical class 17

Topic: Antibiotics. Nitroimidazoles and nitrofurans. fluoroquinolones. Linezolid. Sulfonamides. Trimethoprim.TB.

Maximum points – 2.5

Learning Outcomes:

1. *Explain the difference between antibiotics and synthetic antituberculous drugs*
2. *Compare different sulfonamide drugs*
3. *Explain mechanisms of action of combined drugs (trimethoprim)*
4. *Characterize (indications, contraindications, side effects) these drugs: metronidazole, furazolidone, nitroksolin, ciprofloxacin, linezolid, Sulfametoksazol, trimethoprim, isoniazid, pyrazinamide, ethambutol, rifampicin, ethionamide, streptomycin, PASA*

Practical class 18

Topic: Antiviral drugs. Treatment of HIV infection. Antifungals

Maximum points – 2.5

Learning Outcomes:

*1. Explain mechanisms of action of antiviral drugs.*

*2. Compare the different drugs against HIV.*

*3. Define antifungal for dermatomycosis and systemic mycosis.*

*4. Characterize (indications, contraindications, side effects) these drugs: acyclovir, rimantadine, ribavirin, sofosbuvir, interferons, amphotericin B, ketoconazole, fluconazole, caspofungin*

**METHODICAL INSTRUCTIONS FOR PRACTICAL CLASSES**

Aim: This course is an introduction to pharmacology based on evidence-based medicine and placebo-controlled clinical trials. The course gives students a basic understanding of modern pharmacology and gives a broad overview of the relationship between the basic concepts in general biology (including cell transport, biochemistry and metabolism) and the drugs that affect them. The principles and mechanisms of the action of drugs in a clinical context, as well as at the cellular level are considered, then this knowledge is integrated into a single system. The concepts of anatomy, molecular biology and physiology are illustrated by medical examples to engage students in analytical thinking and to stimulate independent as well as joint work on educational material.

Learning outcomes:

1. *Discuss the principles of modern pharmacology based on the current achievements of science;*
2. *write a prescription for a drug;*
3. *apply the principles of pharmacokinetics in the work (absorption, distribution, biotransformation and excretion of drugs;*
4. *apply the principles of pharmacodynamics (mechanisms of action of drugs at the molecular level);*
5. *list about the main groups of drugs, their mechanisms of action, indications and contraindications for their use.*
6. *describe of unwanted and adverse drug reactions,*
7. *use the principles of evidence-based pharmacology and evidence-based medicine, justify the use of a drug from the perspective of evidence-based medicine;*
8. *apply the basics of medical international terminology, from the field of pharmacology;*
9. *integrate knowledge of anatomy, physiology and biochemistry to explain the mechanisms of action of drugs;*
10. *independently find, analyze and summarize educational and scientific information in relation to situations related to the course content;*

Work schedule

1. Familiarize yourself with the basic and additional literature, use textbooks, the syllabus and present directions, Internet resources to prepare for seminars.

2. Be prepared for class and participate actively on case-discussion and problem solving group activities.

3. Use the examples (in this number cases and your own experience studied before) for illustration of theoretic material.

4. Use different tools for studying, discussion and visualisation of thoughts - drawing, mind maps, 3d-modelling.

5. Use the group work with cases for the development of teamwork skills, communication, problem solving and self-studying.

Scale of response quality

| Evaluation | Criteria | Scale,  points |
| --- | --- | --- |
| Excellent | 1. All key aspects included and presented logically;  2. High accuracy (relevance, without redundancy) and consistent focus on question;  3. Excellent integration of theoretical issues;  4. Provision of relevant examples;  5. In-depth analysis and theoretical justification of given problem (if applicable), all key aspects identified and interpreted;  6. Fluency in use of professional terminology | 90 - 100 |
| Good | 1. All key aspects included and presented logically;  2. Consistent focus on question with satisfactory accuracy, and relevance, and/or some redundancy;  3. Satisfactory integration of theoretical issues;  4. Lack of examples;  5. Satisfactory analysis and theoretical justification of given problem (if applicable), most key aspects identified and interpreted;  6. Correct use of professional terminology | 75 - 89 |
| Satisfactory | 1. Most key aspects included;  2. Satisfactory focus on question - some lapses of relevance and/or noticeably redundancy;  3. Theoretical issues presented without noticeably integration;  4. Provision of unsuccessful examples or no examples;  5. Some analysis and theoretical justification of given problem (if applicable), most key aspects identified and interpreted;  6. Correct use of professional terminology | 50 - 70 |
| Unsatisfactory (FX) | 1. Most key aspects missed;  2. Lack of focus on question - no relevance and notable redundancy;  3. Some theoretical issues presented in someway;  4. No or irrelevant examples;  5. Some analysis and theoretical justification of a given problem (if applicable), most key aspects missed;  6. Lapses in use of professional terminology | 25 - 49 |
| Failed | 1. Most or all key aspects missed;  2. No focus on question, irrelevant information;  3. Theoretical issues missed or superficial;  4. No or irrelevant examples;  5. No analysis and no theoretical justification of a given problem (if applicable), most key aspects missed;  6. Lapses in use of professional terminology | 0-24 |

TEAMWORK GUIDELINES

The medical profession involves working in multidisciplinary teams, so these skills are identified as key in the competence of the doctor and other health professionals in all countries.

Therefore, group work is included as an essential component in the practical exercises of our course. In addition, it aims to provide a safe environment in which you can try out new ideas and practices and acquire relevant group skills. These can be tasks for performance in pairs, triples or small groups of 4-6 people (work with cases, tasks of the ISW, etc.).

When you are working on a project or task in a team, you have the opportunity to use the various strengths of the group members to create a wider and better project or task than if you were working independently.

Group training means you need to share your knowledge and ideas with other students. There are two benefits to this: you need to think carefully about your own ideas in order to explain them to others, and you expand your own understanding, taking into account the knowledge and ideas of others.

Interpersonal Communication and Discussion

Take some time to chat and get to know each of your group mates. The better you know each other and the more convenient you communicate, the more effective you can work together.

Create a culture of mutual respect in your group. You probably had little choice or no choice at all when forming training groups and small teams in the classroom. Therefore, you will have to learn to overcome the differences between people. In addition, you will not have the opportunity to choose employees in the workplace, and at work, you will experience much greater pressure to be a productive member of the team.

For effective communication and discussion in a team: you should not be shy to express your opinion and it is important to feel that these opinions will be heard; it is necessary to feel that all members of the group make a feasible contribution to solving problems, observing agreed rules and plans, performing work efficiently and on time; it is important to know that everyone’s feelings are taken into account by team members, but the goals and objectives of the group are not compromised, in favor of the whims or desires of individual members;

Try to express your opinion and listen to others. There is nothing wrong with disagreeing with your classmates, no matter how confident they are. When you disagree, be constructive and focus on the problem, not the person. Similarly, when someone disagrees with you, respect what he says and the risk that he takes upon himself to express his opinion. Try to find a way that everyone can agree with, and this is not necessarily the opinion of the loudest or smartest member of the team. Below we provide some examples of constructive and destructive group behavior[[1]](#footnote-0):

Constructive group behavior - a person who:

*Unites* - interest in the views and opinions of others and willingness to adapt to interest

*Clarifies* - clearly defines the problems for the group by listening, summarizing, focusing the discussion

*Inspires* - encourages the group, stimulates participation and progress

*Harmonizes* - stimulates group unity and teamwork. For example, uses humor as a relaxation after difficult situations.

*Take the risk* - willingness to take risks at the expense of oneself for the success of the group or project

*Manages the process* - organizes a group on the issues of the process: for example, plan, schedule, timeline, topic, solution methods, and use of information

Destructive group behavior:

*Domination* - takes a lot of time expressing your opinion and views. Trying to take control by capturing energy, time, etc.

*Fussiness* - hastens the group to move quickly before the task is completed. Impatient in listening to other opinions and working together.

*Suspension* - removes itself from a discussion or decision. Opt out

*Ignoring* - does not respect or belittle the ideas and suggestions of the team or individuals. An extreme manifestation of ignoring is an insult in the form of ridicule.

*Distraction* - excessive talkativeness, tells stories and leads groups away from the goal

*Blocking* - prevents group progress by denying all ideas and suggestions. “It will not work because ...”

Effective group work does not arise by itself. A conscious and planned effort is needed, and since many people participate in it, one cannot rely on memory; need to make notes. The following steps will help you and your team work together effectively.

*1. Define clear objectives.* At each stage, you should try to coordinate the tasks. They include a timeline for the project, as well as more specific tasks (such as “agree on an approach to the task before Friday”). Each meeting or discussion should also begin with a specific goal (for example, make a list of tasks that need to be completed). Tasks should be broken down into smaller parts and planned. Sometimes one part cannot be started until the other part is finished, so you may need to draw a simple temporary map.

· discuss the resources that you have and those that you will need to find.

· formulate the desired result.

· consider how you know when you did it well enough?

· split tasks between the team and

· set deadlines for subtasks and time for future meetings.

*2. Set the basic rules.* Discussions can become erratic and can prevent more modest group members from participating if you do not have rules to stimulate discussion, resolve disagreements, and make decisions without repetition. Set the rules from the start and change them as needed. For example: an interesting rule that was developed by one group - anyone who missed a meeting would buy the rest of the group coffee in a coffee shop. No one ever missed a meeting after that.

*3. Communicate effectively.* Make sure you regularly communicate with group members. Try to be clear and positive in what you say without repeating.

*4. Find consensus.* People work together most effectively when they work towards a goal with which they have agreed. Make sure everyone has their own opinion, even if you need time to get more participants to say something. Make sure you listen to everyone’s ideas and then try to come to an agreement that everyone shares and everyone has contributed.

*5. Define the roles.* Divide the work that needs to be done into separate tasks, for which you can use the strengths of individual team members. Define roles for both fulfilling your tasks and for meetings / discussions (for example, Arani is responsible for summarizing the discussions, Joseph is for everyone to express their opinions and make decisions, etc.).

Examples of roles and functions:

*Facilitator* or *leader* (depending on context) - to clarify the goals of the meeting and to summarize the discussions and decisions; ensures that the meeting takes place, continues and the basic rules are respected.

*Secretary* - keep a record of the ideas discussed and decisions made and who does what.

*Time Manager* - to make sure that you discuss everything that you need in the time allotted for the meeting.

*Controller* - to ensure that work is completed by an agreed time, and to solve problems if they are not being performed.

*A process observer* is someone who monitors the process, not the content, and can bring problems to the attention of the team. In this role, it is important to be positive, not condemning.

*Editor* - bring all materials together, identify gaps or matches and ensure consistency in the final presentation.

*6. Make it clear.* When a decision is made, it should be explained in such a way that it is absolutely clear to everyone that it was decided, including the time frame.

*7. Keep good notes.* Always summarize the discussions and document the decisions and publish them (for example in WhatsApp chat) so you can always get back to them. This includes lists of those who agreed what to do.

*8. Stick to the plan.* If you agreed to do something as part of the plan, do it. Your group relies on you to do what you agreed to do, and exactly in this way, not in the way you would like. If you think the plan should be reviewed, discuss it.

*9. Keep track of progress and keep up to date*. Discuss progress together regarding your schedule and deadlines. Make sure you meet deadlines personally so you do not let your group down.

Co-writing a document / report

Joint writing is one of the most difficult parts of group work. There are many ways to do this, and your group must decide how to separate the work of writing, comparing, editing, and finalizing your work. Writing in a group (six people crowd around the keyboard) is a recipe for conflict and lack of progress. The other extreme - when one person assumes all responsibility and ultimately does most of the work - is also unproductive and contributes to conflict.

Three approaches are possible when working on a common document:

1 - One person writes the most part - this means that a narrow circle of ideas is used, and the rest of the team does not learn (and will not learn) to write reports and documents.

2 - Each person writes one section - then it is difficult to make a single consistent report, and you will not know about the rest, except for your own section.

3- Co-writing. This is the most productive way to solve group problems and provides the greatest benefit from collaboration. For example: in each section, there is a writer and at least one reviewer, and each team member is the author of a section and a reviewer of another one.

All team members before finalization by the editor must review the final product. Alternatively, you can have one author with others, editors, add and review, and someone tidies the finished report.

Try to divide the writing of source documents into tasks and solve them individually or in pairs. After the first draft of the sections are written, send out all the components and read them. You will probably need to come together to discuss how to combine them so that they fit together. Any participants who were not involved in preparing the drafts can do part of this work. Then edit, improve and polish the draft. It’s convenient to collaborate on documents in Google documents.

When preparing a report / final document, regularly check the following:

- Is the purpose of the project clear from the report?

- Are the conclusions or recommendations clear?

- Do conclusions follow from the main part of the report?

- Do sections fit well?

- Does the report achieve goals (and evaluation criteria)?

- Are the necessary components sufficiently covered?

Whatever method you use, all group members must agree on the process and how they are going to maximize the collaborative approach to writing the final document.

Monitoring team performance and coping

Below is a checklist that includes a list of common problems that arise in a group work. Use it regularly to identify problems before they get out of hand. If serious problems and tensions arise, use it to determine where something might go wrong. First answer each question about yourself, and then give answer to this question about the group as a whole. Then gather a group and discuss where, in your opinion, problems may arise, and think about how you can overcome these problems.

Each participant must complete this checklist. You should do this exercise regularly to track and improve your team’s performance.

1. Answer each question regarding your teamwork.

2. Answer each question regarding the rest of the team.

3. Get together with your entire team and discuss where, in your opinion, any problems arise.

4. Discuss what you are going to do to overcome these problems.

Checklist for self-assessment of team effectiveness.

| You | Me personally | Group as a whole | Comments |
| --- | --- | --- | --- |
| Effectively clarify your tasks and tasks at each stage? |  |  |  |
| Evaluate the progress of work? |  |  |  |
| We clarify and document everything that the group decided? |  |  |  |
| We clarify who will do what and how? |  |  |  |
| We clarify by what date each task should be done? |  |  |  |
| Setting meeting management rules? |  |  |  |
| Adhere to agreed rules? |  |  |  |
| Listening to each other? |  |  |  |
| Allow some team members to dominate? |  |  |  |
| Allow some team members to refuse / withdraw? |  |  |  |
| We sacrifice personal desires for the success of the team? |  |  |  |
| Recognize the feelings of other team members? |  |  |  |
| Making equal contributions to team progress? |  |  |  |
| Adhere to agreed rules for writing and naming files? |  |  |  |

Points and Grade

Group tasks and assignments mean that grades are given to the whole group based on the results of the work of the whole group. Everyone should be interested in ensuring the effective contribution of all members of the group and ensuring the high quality of the assignment. Sometimes, to assess the relative contribution of each to the group process, a form of peer-to-peer or peer review and a team assessment form will be used. This can be used to moderate assignment grades, or simply as a way to give feedback on your work in a group. The following are examples of student assessment criteria for team training.

| № | Student assessment criteria in practical classes |
| --- | --- |
| 1 | *Preparation for classes:*  He studies information focused on the case and problematic issues, uses various sources, and supports the statements with relevant links. |
| 2 | *Group skills and professional attitude:*  Demonstrates excellent attendance, reliability, responsibility Takes the initiative, takes an active part in the discussion, helps the teammates, willingly takes on tasks |
| 3 | *Communication skills:*  Actively listens, shows emotions according to the situation, is susceptible to non-verbal and emotional signals, shows respect and correctness in relation to others, helps to resolve misunderstandings and conflicts |
| 4 | *Feedback Skills:*  Demonstrates a high level of introspection, critically evaluates oneself and colleagues, provides constructive and objective feedback in a friendly manner, accepts feedback without opposition |
| 5 | *Skills of critical thinking and effective learning:*  Effectively participates in generating hypotheses and formulating problematic questions, gives relevant examples from life, skillfully applies knowledge to the problem / case under consideration, critically evaluates information, draws conclusions, explains and substantiates statements, draws diagrams and drawings, demonstrates a constant interest in the material being studied |
| 6 | *Theoretical knowledge and skills on the topic of the lesson:*  All key aspects are presented logically; accuracy, relevance of answers to the questions posed without redundancy; integration of theoretical issues; Use of relevant examples proper use of professional terminology |

Basic literature:

1. Thompson & Thompson genetics in medicine (2016) Robert L. Nussbaum, Roderick R. McInnes, Huntington F. Willard, Ada Hamosh. [Philadelphia, PA: Elsevier](http://cat.lib.unimelb.edu.au/search~S30?/hElsevier%2C/helsevier/-3,-1,0,B/browse)
2. Basic & Clinical Pharmacology [Electronic resource]: collection / ed.: B. G. Katzung, A. J. Trevor. - 13th ed. - New York; Chicago; San Francisco: McGraw-Hill Education, 2015. - 1837 p. - ISBN 978-0-07-182641-9: 0.00
3. Jawetz, Melnick & Adelberg’s Medical microbiology. Geo F. Brooks, Karen C. Carroll, Janet S. Butel, Stephen A. Morse, Timothy A. Mietzner. 26th edition, 2013

Additional literature:

1. Jorde, L.B. et al. (2016) Medical Genetics. [Philadelphia, PA: Elsevier](http://cat.lib.unimelb.edu.au/search~S30?/hElsevier%2C/helsevier/-3,-1,0,B/browse)
2. Emery’s Elements of Medical Genetics (2017) Turnpenny, P.D., Ellard S. 15th Edition, Elsevier
3. Hartwell, L. et al (2017) Genetics: from genes to genomes, 6th edition. New York, NY: McGrawHill Education
4. USMLE Step 1 Lecture Notes (2017): Biochemistry and Medical Genetics. [Kaplan Publishing](https://www.bookdepository.com/publishers/Kaplan-Publishing)

WWW resources:

1. OMIM® Online Mendelian Inheritance in Man® An Online Catalog of Human Genes and Genetic Disorders <https://www.omim.org/>
2. The Genetic Testing Registry (GTR®) <https://www.ncbi.nlm.nih.gov/gtr/>
3. Genetics Home Reference. <https://ghr.nlm.nih.gov/resources>
4. ClinGen: Clinical Genome Resource <https://www.clinicalgenome.org/>
5. Learn.Genetics <https://learn.genetics.utah.edu/content/basics/>
6. Clinical Genetic Education Resources (Courses and Lectures) <https://www.kumc.edu/gec/prof/genecour.html>
7. Genomics Education Program. [https://www.genomicseducation.hee.nhs.uk](https://www.genomicseducation.hee.nhs.uk/education/)
8. ELSEVIER “Clinical learning” training program, 2018
9. <https://www.msdmanuals.com/professional/clinical-pharmacology>

1. adapted from Brunt (1993):<https://tle.wisc.edu/solutions/engagement/constructive-and-destructive-groupbehaviors>

   [↑](#footnote-ref-0)